CONDITION

Glioma-polyposis syndrome

Glioma-polyposis syndrome is an inherited condition linked to bowel polyps and certain brain tumors.

WHAT IT IS
Inherited tendency to bowel polyps and brain tumors

BODY SYSTEM
Digestive and nervous systems

MAIN SYMPTOMS
Rectal bleeding, bowel changes, headaches, seizures

WHAT CAUSES IT
Inherited APC or DNA mismatch-repair gene changes

HOW IT IS DIAGNOSED
Colonoscopy, brain MRI, tissue testing, and genetic testing

MAIN TREATMENT
Surveillance, surgery, and tumor-specific cancer treatment

CAN IT BE CURED?
Sometimes - tumors may be removed; inherited risk remains

IS IT AN EMERGENCY?
Sometimes - new seizure or sudden neurological symptoms

LONG-TERM OUTLOOK
Lifelong surveillance; outlook shaped by tumor type and stage

Overview

Glioma-polyposis syndrome is a rare inherited condition associated with bowel polyps and certain tumors of the brain or spinal cord. A glioma is a tumor that develops from glial-type cells in the nervous system. Polyposis means that multiple growths called polyps form in the bowel.

Most bowel polyps involved in this condition are adenomas. These are not cancer, but some can develop into colorectal cancer over time. Brain tumors may be cancerous or non-cancerous, although some associated gliomas can grow quickly.

The name does not always refer to one specific genetic disorder. Similar combinations of bowel and brain tumors can occur with:

Mismatch-repair genes help cells correct mistakes made when DNA is copied. When this repair system does not work properly, tumors can form more easily.

Identifying the underlying gene matters because it affects which tumors are more likely, when they may occur, and what surveillance may be appropriate. For example, APC-related polyposis is more strongly associated with medulloblastoma, while mismatch-repair conditions may be associated with gliomas. These patterns are not absolute.

- Changes in the APC gene, which cause APC-associated polyposis
- Changes in DNA mismatch-repair genes, which may cause Lynch syndrome
- Changes in both copies of a mismatch-repair gene, causing constitutional mismatch repair deficiency

Symptoms

Bowel polyps often cause no symptoms. They may be discovered during a colonoscopy arranged because of family history or genetic test results.

When bowel symptoms occur, they may include:

Symptoms from a brain or spinal cord tumor depend on its location, size, and speed of growth. Possible symptoms include headaches that become more frequent, seizures, repeated vomiting, weakness, numbness, balance problems, or changes in speech or vision.

A tumor may also cause changes in memory, concentration, mood, personality, school performance, or everyday abilities. These symptoms have many causes other than a brain tumor, but persistent or worsening changes should be assessed.

In children, signs may be less specific. They can include developmental changes, loss of skills, unusual clumsiness, a change in walking, or a noticeable decline at school.

- Blood in or on the stool
- A lasting change in bowel habits
- Abdominal discomfort
- Unexplained iron-deficiency anemia, which can cause tiredness
- Unintentional weight loss

Causes and risk factors

Glioma-polyposis syndrome is usually related to an inherited gene change. A gene change may

also be called a pathogenic variant.

The main genes involved fall into two groups.

The APC gene normally helps control cell growth. A harmful change can cause familial adenomatous polyposis, in which many colorectal adenomas develop, often beginning at a young age. Some people also have tumors outside the bowel.

DNA mismatch-repair genes include MLH1, MSH2, MSH6, PMS2 , and EPCAM , which can affect MSH2. One inherited altered copy may cause Lynch syndrome. Lynch syndrome usually causes fewer polyps than classic APC-related polyposis, but it raises the risk of colorectal and several other cancers.

A child who inherits harmful changes in both copies of a mismatch-repair gene can have constitutional mismatch repair deficiency. This condition can cause bowel polyps, brain tumors, blood cancers, and other cancers during childhood or early adulthood.

How the condition is inherited therefore depends on the gene involved. In some families, one altered gene copy is enough to pass on an increased risk. Constitutional mismatch repair deficiency usually requires an altered copy from each parent.

A family may appear to have no previous history if the gene change is new, the family is small, relatives have not been tested, or affected relatives died before a diagnosis was made.

Diagnosis

There is no single test that identifies every form of glioma-polyposis syndrome. Diagnosis brings together the personal and family history, bowel examination, brain imaging, tumor findings, and genetic testing.

A colonoscopy uses a flexible camera to examine the colon and rectum. Polyps can be removed or sampled during the procedure. A pathologist then examines the tissue under a microscope to determine the type of polyp and whether cancer is present.

Magnetic resonance imaging, or MRI, creates detailed pictures of the brain or spinal cord. If imaging finds a tumor, surgery or a biopsy may be needed to identify its exact type and molecular features. A biopsy removes a small tissue sample for laboratory examination.

Tumor tissue may be tested for signs that DNA mismatch repair is not working. Tests include mismatch-repair protein staining and microsatellite instability testing. These findings can guide genetic investigation, but they do not always prove that a gene change was inherited.

A germline genetic test looks for a gene change present throughout the body. Testing is usually arranged with genetic counselling because results can affect blood relatives as well as

the person being tested. Sometimes testing does not find a clear cause even when the clinical history strongly suggests an inherited condition.

Treatment and management

Management is based on the underlying gene, the number and type of bowel polyps, and whether a nervous system tumor or other cancer is present. Care may involve genetics, gastroenterology, colorectal surgery, neurology, neurosurgery, and oncology teams.

Regular colonoscopy can find and remove some polyps before they become cancerous. If there are too many polyps to manage safely through colonoscopy, or if cancer is found, surgery to remove part or all of the colon may be considered. The exact operation depends on the rectal polyp burden, cancer findings, age, and the underlying condition.

Surveillance may also include other organs. The recommended tests differ between APC-associated polyposis, Lynch syndrome, and constitutional mismatch repair deficiency. Routine brain MRI is not handled the same way in every inherited syndrome, so the plan is based on the confirmed genetic diagnosis and family history.

Treatment for a glioma may include:

The tumor's grade describes how abnormal its cells look and how it is expected to behave. Its molecular features can also influence treatment. Some mismatch-repair-deficient cancers may be considered for immunotherapy, which helps the immune system recognize cancer cells, but suitability depends on the individual tumor and clinical situation.

Treatment can address existing polyps and tumors. It does not remove an inherited gene change, so long-term surveillance remains important.

- Surgery to remove as much tumor as safely possible
- Radiotherapy, which uses targeted radiation
- Chemotherapy or other medicines
- Rehabilitation for problems with movement, speech, or daily activities
- Medicines to control seizures, swelling, pain, or nausea

Complications

Bowel adenomas can become colorectal cancer if they are not found and managed. Polyps or tumors may also bleed, causing anemia.

A brain tumor can affect movement, vision, speech, memory, behavior, or seizure control. Some tumors may return or continue growing after treatment. Surgery, radiotherapy, and medicines can also have lasting effects, including fatigue, bowel changes, fertility problems, or difficulties with learning and concentration.

Depending on the gene involved, there may be increased risks of tumors elsewhere in the digestive system or in organs such as the uterus, ovaries, urinary tract, or skin. Constitutional mismatch repair deficiency can also raise the risk of blood cancers.

Prevention

An inherited gene change cannot currently be prevented through diet, exercise, or other lifestyle choices. There is also no established way to prevent every associated brain tumor.

However, surveillance can reduce the chance that bowel polyps progress unnoticed. Removing polyps and, when needed, having preventive bowel surgery can substantially reduce colorectal cancer risk. These measures do not eliminate all cancer risk.

Once a familial gene change has been identified, adult relatives can choose genetic counselling and testing. People planning a pregnancy may also discuss reproductive testing options. These are personal choices, not requirements.

Living with it and what to expect

Glioma-polyposis syndrome usually requires lifelong follow-up, even after polyps or a tumor have been treated. The surveillance plan may change with age, new test results, the number of polyps, and family history.

Some people remain well for long periods with regular screening and timely polyp removal. Others need bowel surgery or treatment for one or more tumors. If the colon is removed, bowel habits may change permanently, but many people return to work, education, travel, and other usual activities after recovery.

The outlook is shaped mainly by the underlying genetic condition and the type, location, grade, and stage of any tumor. A slow-growing, removable tumor has a different course from an aggressive glioma or cancer that has spread. No single course applies to everyone with this label.

Ongoing care may include rehabilitation, seizure management, psychological support, and help with school or work. Genetic results can also bring uncertainty or concern for relatives, and genetic counselling can help families understand what the findings do and do not mean.

When to get medical help

Arrange a routine medical assessment for persistent bowel bleeding, a lasting change in bowel habits, unexplained anemia, repeated headaches, new balance problems, or gradual changes in memory, behavior, speech, or vision.

Medical review is also appropriate if close relatives have had numerous bowel polyps, colorectal cancer at a young age, brain tumors, or a known APC or mismatch-repair gene change. This does not mean that you have the condition, but it may justify a detailed family-history assessment.

Seek urgent or emergency help for:

These symptoms can have causes other than glioma-polyposis syndrome, but they need prompt assessment.

- A first seizure or a seizure lasting longer than usual
- Sudden weakness, facial drooping, speech difficulty, or loss of vision
- Loss of consciousness or severe confusion
- A sudden, extremely severe headache
- Repeated vomiting with worsening headache or drowsiness
- Heavy rectal bleeding, black stools, fainting, or marked dizziness

Common questions

Is glioma-polyposis syndrome the same as bowel cancer?

No. Polyposis means that multiple bowel polyps are present. Most adenomatous polyps are not cancer when they first form, but some can become cancer over time. Surveillance and polyp removal are intended to reduce that risk.

Does having bowel polyps mean I have a brain tumor?

No. Bowel polyps are common and usually are not linked to a brain tumor syndrome. Concern is greater when there are many adenomas, polyps begin unusually early, or the family has a pattern of bowel and brain tumors.

Can a brain tumor be the first sign?

Yes. In some people, a brain tumor is identified before bowel polyps or colorectal cancer. In others, polyposis is discovered first. The order can be influenced by the underlying gene and the person's age.

Will my children inherit it?

That depends on the gene. Some related conditions can be passed on when one parent carries an altered gene copy. Constitutional mismatch repair deficiency usually occurs when a child inherits an altered mismatch-repair gene copy from each parent. Genetic testing can clarify the inheritance pattern in a family.

Can removing the colon cure the condition?

Removing the colon can greatly reduce colorectal cancer risk from that organ, but it does not remove the inherited gene change. Surveillance may still be needed for the remaining rectum, surgically created bowel pouch, upper digestive tract, nervous system, or other organs.

Do all gliomas linked to this condition have the same outlook?

No. Gliomas differ in location, grade, molecular features, and response to treatment. Age and general health also matter. The pathology and molecular test results from the tumor provide more useful information than the syndrome name alone.

Sources

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